



SLC7A9 gene

solute carrier family 7 member 9

Normal Function

The *SLC7A9* gene provides instructions for producing one part (subunit) of a protein made primarily in the kidneys. This subunit joins with another protein subunit, produced from the *SLC3A1* gene, to form a transporter protein complex. During the process of urine formation in the kidneys, this protein complex absorbs particular protein building blocks (amino acids) back into the blood. In particular, the amino acids cystine, ornithine, arginine, and lysine are absorbed back into the blood through this mechanism.

Health Conditions Related to Genetic Changes

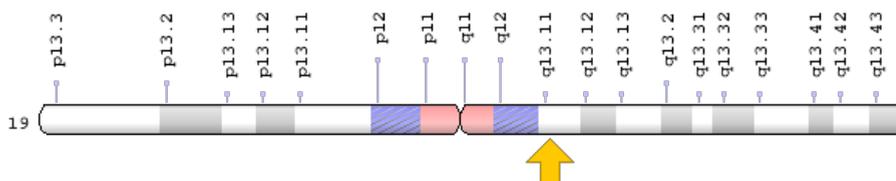
cystinuria

At least 95 mutations in the *SLC7A9* gene have been found to cause cystinuria. Many of these mutations alter a single DNA building block (nucleotide) or insert or delete a small number of nucleotides in the *SLC7A9* gene. These changes lead to an abnormally functioning transporter protein complex, which causes certain amino acids to become concentrated in the urine. Cystine is the only amino acid that forms crystals and stones in the bladder or kidneys, leading to the signs and symptoms of cystinuria.

Chromosomal Location

Cytogenetic Location: 19q13.11, which is the long (q) arm of chromosome 19 at position 13.11

Molecular Location: base pairs 32,830,511 to 32,870,958 on chromosome 19 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- b0,+AT
- BAT1_HUMAN
- CSNU3
- solute carrier family 7 (amino acid transporter light chain, bo,+ system), member 9
- solute carrier family 7 (glycoprotein-associated amino acid transporter light chain, bo,+ system), member 9
- solute carrier family 7, member 9

Additional Information & Resources

Educational Resources

- National Institute of Diabetes and Digestive and Kidney Diseases: Your Kidneys and How They Work
<https://www.niddk.nih.gov/health-information/kidney-disease/kidneys-how-they-work>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28SLC7A9%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- SOLUTE CARRIER FAMILY 7 (CATIONIC AMINO ACID TRANSPORTER, y+ SYSTEM), MEMBER 9
<http://omim.org/entry/604144>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_SLC7A9.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=SLC7A9%5Bgene%5D>
- HGNC Gene Family: Solute carriers
<http://www.genenames.org/cgi-bin/genefamilies/set/752>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=11067

- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/11136>
- UniProt
<http://www.uniprot.org/uniprot/P82251>

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<https://ghr.nlm.nih.gov/gene/SLC7A9>

Reviewed: January 2009
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services